

We Claim:

1. An isolated polynucleotide encoding equine laminin  $\gamma$ 2.
2. An isolated polynucleotide as defined in claim 1, which is RNA.
3. An isolated polynucleotide as defined in claim 1, which is DNA.
4. An isolated polynucleotide as defined in claim 1, comprising the nucleic acid sequence of nucleotides 1-3570 of SEQ ID No:1.
5. An isolated polynucleotide as defined in claim 1, which encodes laminin  $\gamma$ 2 having the amino acid sequence of 1-1190 of SEQ ID No:2.
6. Equine laminin  $\gamma$ 2, in a form essentially free from other proteins of mammalian origin.
7. Equine laminin  $\gamma$ 2, having the amino acid sequence of SEQ ID No:2.
8. Equine laminin  $\gamma$ 2, that is encoded by a polynucleotide having the nucleotide sequence of nucleotides 1-3570 of SEQ ID No:1 .
9. A recombinant DNA construct incorporating the polynucleotide of claim 1.
10. A cell having incorporated expressibly therein a construct as defined in claim 9.
11. A process for obtaining a substantially homogeneous source of equine laminin  $\gamma$ 2, comprising the steps of culturing cells having incorporated expressibly therein a polynucleotide as defined in claim 1, and then recovering the equine laminin  $\gamma$ 2 therefrom.
12. A method of diagnosing epidermolysis bullosa in a horse comprising the steps of:
  - 1) obtaining a biological sample from the horse;
  - 2) isolating nucleic acid therefrom and amplifying laminin  $\gamma$ 2-encoding nucleic acid using appropriate primers; and
  - 3) analysing the amplified nucleic acid to identify the presence of mutated laminin  $\gamma$ 2-encoding nucleic acid having a cytosine insert at position 1368, wherein the homozygous presence of said mutated laminin  $\gamma$ 2-encoding nucleic acid indicates a diagnosis of epidermolysis bullosa.
13. A method as defined in claim 12, wherein the primers used to amplify the laminin  $\gamma$ 2-encoding nucleic acid were (sense) 5'-TGTTACTCAGGGATGAGAA-3' (SEQ ID No: 29) and (antisense) 5'-CTGGGGGCAGTTATTGCAC-3' (SEQ ID No: 30).

14. A method as defined in claim 12, wherein the amplified nucleic acid is chromatographically analysed to identify the heterozygous presence of the mutated laminin  $\gamma$ 2-encoding nucleic acid.
15. A kit for diagnosing epidermolysis bullosa in horses comprising the nucleic acid primers 5'-TGTTACTCAGGGATGAGAA-3' (SEQ ID No: 29) and (antisense) 5'-CTGGGGGCAGTTATTGCAC-3' (SEQ ID No: 30).
16. A monoclonal or polyclonal antibody directed against equine laminin  $\gamma$ 2.
17. A method of diagnosing JEB in a horse comprising:
- 1) obtaining a biological sample from a horse;
  - 2) isolating the protein component from the sample; and
  - 3) screening the sample for laminin  $\gamma$ 2 peptide, wherein absence of laminin  $\gamma$ 2 peptide in the sample is indicative of JEB.
18. A method as defined in claim 17, wherein the sample is screened with an antibody directed against equine laminin  $\gamma$ 2.
19. A method as defined in claim 12, wherein the sample is obtained from an unborn foal.
20. A method as defined in claim 17, wherein the sample is obtained from an unborn foal.

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1. An isolated polynucleotide encoding equine laminin  $\gamma$ 2.
2. An isolated polynucleotide as defined in claim 1, which is RNA.
3. An isolated polynucleotide as defined in claim 1, which is DNA.
4. An isolated polynucleotide as defined in claim 1, comprising the nucleic acid sequence of nucleotides 1-3570 of SEQ ID No:1.
5. An isolated polynucleotide as defined in claim 1, which encodes laminin  $\gamma$ 2 having the amino acid sequence of 1-1190 of SEQ ID No:2.
6. Equine laminin  $\gamma$ 2, in a form essentially free from other proteins of mammalian origin.
7. Equine laminin  $\gamma$ 2, having the amino acid sequence of SEQ ID No:2.
8. Equine laminin  $\gamma$ 2, that is encoded by a polynucleotide having the nucleotide sequence of nucleotides 1-3570 of SEQ ID No:1 .
9. A recombinant DNA construct incorporating the polynucleotide of claim 1.
10. A cell having incorporated expressibly therein a construct as defined in claim 9.
11. A process for obtaining a substantially homogeneous source of equine laminin  $\gamma$ 2, comprising the steps of culturing cells having incorporated expressibly therein a polynucleotide as defined in claim 1, and then recovering the equine laminin  $\gamma$ 2 therefrom.
- (12) A method of diagnosing epidermolysis bullosa in a horse comprising the steps of:
  - 1) obtaining a biological sample from the horse;
  - 2) isolating nucleic acid therefrom and amplifying laminin  $\gamma$ 2-encoding nucleic acid using appropriate primers; and
  - 3) analysing the amplified nucleic acid to identify the presence of mutated laminin  $\gamma$ 2-encoding nucleic acid having a cytosine insert at position 1368, wherein the homozygous presence of said mutated laminin  $\gamma$ 2-encoding nucleic acid indicates a diagnosis of epidermolysis bullosa.
- (13) A method as defined in claim 12, wherein the primers used to amplify the laminin  $\gamma$ 2-encoding nucleic acid were (sense) 5'-TGTTACTCAGGGGATGAGAA-3' (SEQ ID No: 29) and (antisense) 5'-CTGGGGGCAGTTATTGCAC-3' (SEQ ID No: 30).

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3514?  
12402

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Laminin  $\gamma$ 2  
1368

SEQ 1 =  
laminin  $\gamma$ 2  
1-3570-  
418aa,

20140701-20140701

14. A method as defined in claim 12, wherein the amplified nucleic acid is chromatographically analysed to identify the heterozygous presence of the mutated laminin  $\gamma 2$ -encoding nucleic acid.
15. A kit for diagnosing epidermolysis bullosa in horses comprising the nucleic acid primers 5'-TGTTACTCAGGGATGAGAA-3' (SEQ ID No: 29) and (antisense) 5'-CTGGGGCAGTTATTGCAC-3' (SEQ ID No: 30).
16. A monoclonal or polyclonal antibody directed against equine laminin  $\gamma 2$ .
17. A method of diagnosing JEB in a horse comprising:
- 1) obtaining a biological sample from a horse;
  - 2) isolating the protein component from the sample; and
  - 3) screening the sample for laminin  $\gamma 2$  peptide, wherein absence of laminin  $\gamma 2$  peptide in the sample is indicative of JEB.
18. A method as defined in claim 17, wherein the sample is screened with an antibody directed against equine laminin  $\gamma 2$ .
19. A method as defined in claim 12, wherein the sample is obtained from an unborn foal.
20. A method as defined in claim 17, wherein the sample is obtained from an unborn foal.